



RBPJ gene

recombination signal binding protein for immunoglobulin kappa J region

Normal Function

The *RBPJ* gene provides instructions for making a protein called RBP-J, which is an integral part of a signaling pathway known as the Notch pathway. Notch signaling controls how certain types of cells develop in the growing embryo, including those that form the bones, heart, muscles, nerves, and blood. Signaling through the Notch pathway stimulates the RBP-J protein to attach (bind) to specific regions of DNA and control the activity of genes that play a role in cellular development.

Health Conditions Related to Genetic Changes

Adams-Oliver syndrome

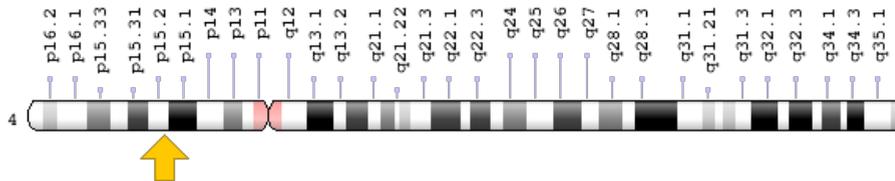
Mutations in the *RBPJ* gene lead to Adams-Oliver syndrome, a condition characterized by areas of missing skin (aplasia cutis congenita), usually on the scalp, and malformations of the hands and feet. These mutations change single protein building blocks in the RBP-J protein, altering the region of the protein that normally binds to DNA. The altered proteins are unable to bind to DNA, preventing the activation of particular genes. These changes in gene activity impair the proper development of the skin on the top of the head and the bones in the hands and feet. It is unclear why impaired development primarily affects these tissues.

rheumatoid arthritis

Chromosomal Location

Cytogenetic Location: 4p15.2, which is the short (p) arm of chromosome 4 at position 15.2

Molecular Location: base pairs 26,163,489 to 26,435,131 on chromosome 4 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- AOS3
- CBF-1
- CBF1
- csl
- H-2K binding factor-2
- IGKJRB
- IGKJRB1
- immunoglobulin kappa J region recombination signal binding protein 1
- KBF2
- RBP-J
- RBP-J kappa
- RBP-JK
- RBPJK
- RBPSUH
- recombining binding protein suppressor of hairless
- recombining binding protein suppressor of hairless isoform 3
- renal carcinoma antigen NY-REN-30
- SUH

- SUH_HUMAN
- suppressor of hairless homolog

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28RBPJ%5BTIAB%5D%29+OR+%28recombination+signal+binding+protein+for+immunoglobulin+kappa+J+region%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- RECOMBINATION SIGNAL-BINDING PROTEIN FOR IMMUNOGLOBULIN KAPPA J REGION
<http://omim.org/entry/147183>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_RBPJ.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=RBPJ%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=5724
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/3516>
- UniProt
<http://www.uniprot.org/uniprot/Q06330>

Sources for This Summary

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<https://ghr.nlm.nih.gov/gene/RBPJ>

Reviewed: November 2015

Published: March 21, 2017

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